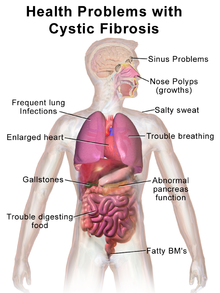
Genetic Counselor: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Period:\_\_\_\_\_\_

**Inherited Genetic Disorder Case Study: Cystic Fibrosis**

Background: Cystic fibrosis, or CF, is one of the most common inherited single gene disorders in Caucasians. About one in 2500 Caucasian babies is born with CF and about one in 25 Caucasians of northern European descent carries the gene for CF. People with CF secrete abnormal body fluids, including unusual sweat and a thick mucus which prevents the body from properly cleansing the lungs. The mucus interrupts the function of vital organs and leads to chronic infections. Classic CF also involves the pancreas and causes decreased absorption of essential nutrients. Life expectancy has improved, but, ultimately, death most often occurs from respiratory failure.

Scenario: Mike and Sarah are planning to have children. They are concerned their child might have cystic fibrosis because Mike’s sister had cystic fibrosis, although neither of his parents had cystic fibrosis. Sarah has no family history of cystic fibrosis.

1. If cystic fibrosis is caused by a recessive allele, which genotype must Mike’s sister have?
2. What must Mike’s parent’s genotypes be if they do not have CF but have a daughter who does? EXPLAIN HOW YOU KNOW!

1. Complete a Punnett Square of a cross between Mike’s parents. (Use F to represent the dominant allele, and f to represent the recessive CF allele)

1. What are the possible genotypes of Mike’s parent’s children and probability of each?
2. What are the possible phenotypes of Mike’s parent’s children and probability of each?

1. Based on the Punnett Square what is the probability that Mike is a carrier for CF?
2. Mike undergoes genetic testing (a DNA sample is obtained through a cheek swab) to determine if he is in fact a carrier for CF. He is. Knowing this and the previous information about Sarah, complete a Punnett Square of their cross.

1. What are the possible genotypes of Mike and Sarah’s children and probability of each?

1. What are the possible phenotypes of Mike and Sarah’s children and probability of each?

1. Can Mike and Sarah have a child with CF? Why or why not?
2. Can Mike and Sarah have a child who is a carrier for CF? Why or why not?

In the past, people with cystic fibrosis typically died in childhood, but modern medicine has allowed men and women with cystic fibrosis to live well past puberty. Therefore, longer lifespans have produced new issues surrounding fertility. Unfortunately, almost all males with cystic fibrosis are infertile. Male infertility is caused by a malformed vas deferens, which is the tube that carries sperm from the testis to the penis. Approximately 98 percent of men with cystic fibrosis are infertile.

Typically, men with cystic fibrosis who want to father a child, require assisted reproduction techniques. This involves removal of the sperm from the male and insertion of the sperm into the male's partner. Unfortunately, men with cystic fibrosis tend to also have malformed sperm and low sperm counts, which complicate this procedure. Moreover, assisted reproduction techniques can be quite expensive and time consuming

Women with cystic fibrosis typically have difficulties with regular menstrual cycles, including missed or irregular periods, or amenorrhea, which is the absence of periods. Typically, irregular cycles are the result of low body weight, poor overall health, and poor control of cystic fibrosis-related diabetes. Women with cystic fibrosis may also have thick mucous that surrounds the cervix, making it difficult for sperm to penetrate into the uterus. Overall, these female infertility issues are relatively well-managed and most women with cystic fibrosis can get pregnant.

1. Complete a Punnett Square of a cross between a woman with CF and a man that neither has CF or is a carrier for CF.

1. What is the likelihood that their child will have CF? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
2. What is the likelihood that their child will be a carrier for CF? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_